# MANAGEMENT OF TRANSITION IN RARE DISEASES - A DELPHI CONSENSUS

**WG**
**Management of transition**

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## Introduction

Child-to-adult transition represents a critical moment in the care of patients with rare diseases. Previous work has unveiled significant differences in clinical practice in this area, urging uniformity across different centers in Europe.[1] Due to the complexity of the process and the large number of variables involved, a literature-based Delphi expert consensus was chosen in order to shed light on the matter.

## Work Plan

To identify recommendations for transitional issues, a scoping literature review on online databases, organisation websites, grey literature and conference abstracts was performed. A multistep web-based Delphi consensus was performed across five different disease groups: Ataxia, HSP, Leukodystrophy, Chorea and Dystonia. Level of impairment in the target population was explored based on ADL.
Final agreement on relevant recommendations was achieved with appropriate questions to panelists.

## Results

The scoping review yielded fifty-nine relevant issues for recommendations. Questionnaires were disseminated to all ERN-RND members and disease-specific specialists. Completed questionnaires were collected from fourty-seven specialists in thirteen countries across five disease groups. A median of thirty-five recommendations met consensus for each disease group. Seventeen recommendations met consensus across all disease groups. ADL impairment showed variablility for different disease groups.

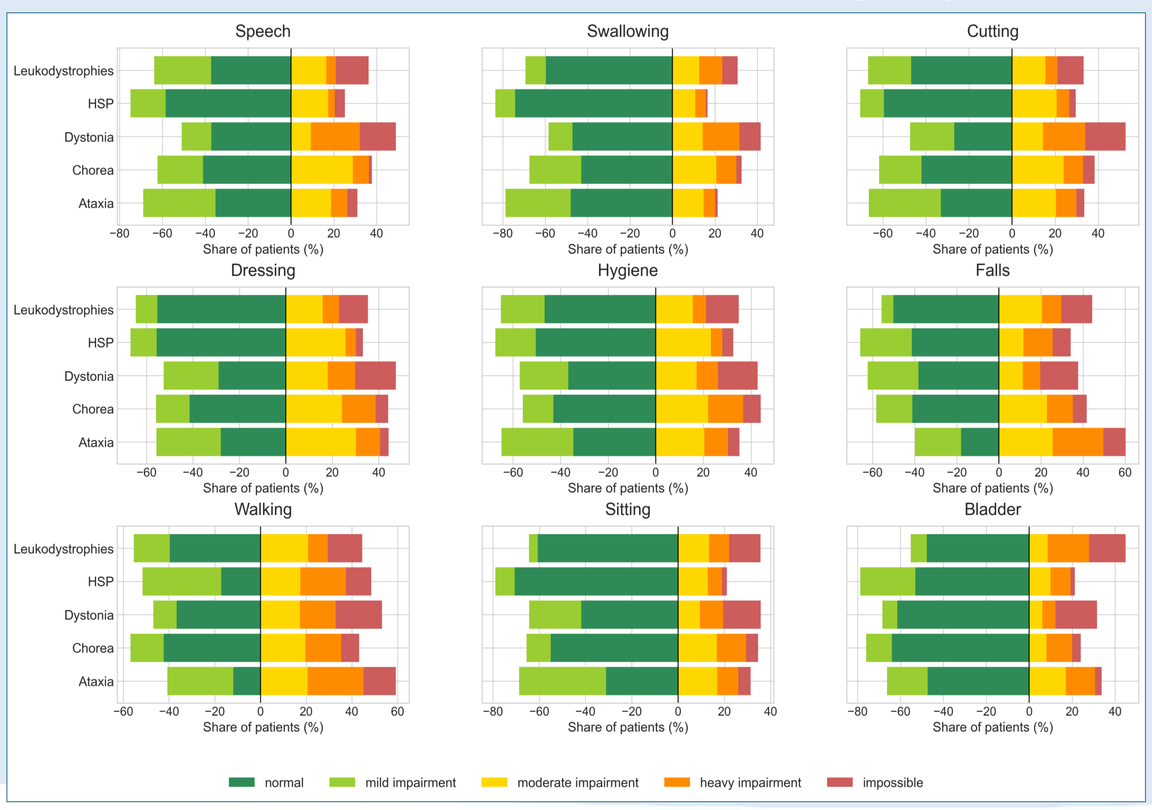


Figure 1.
Levels of ADL impairment across disease groups as reported by panelists referring to their patient populations

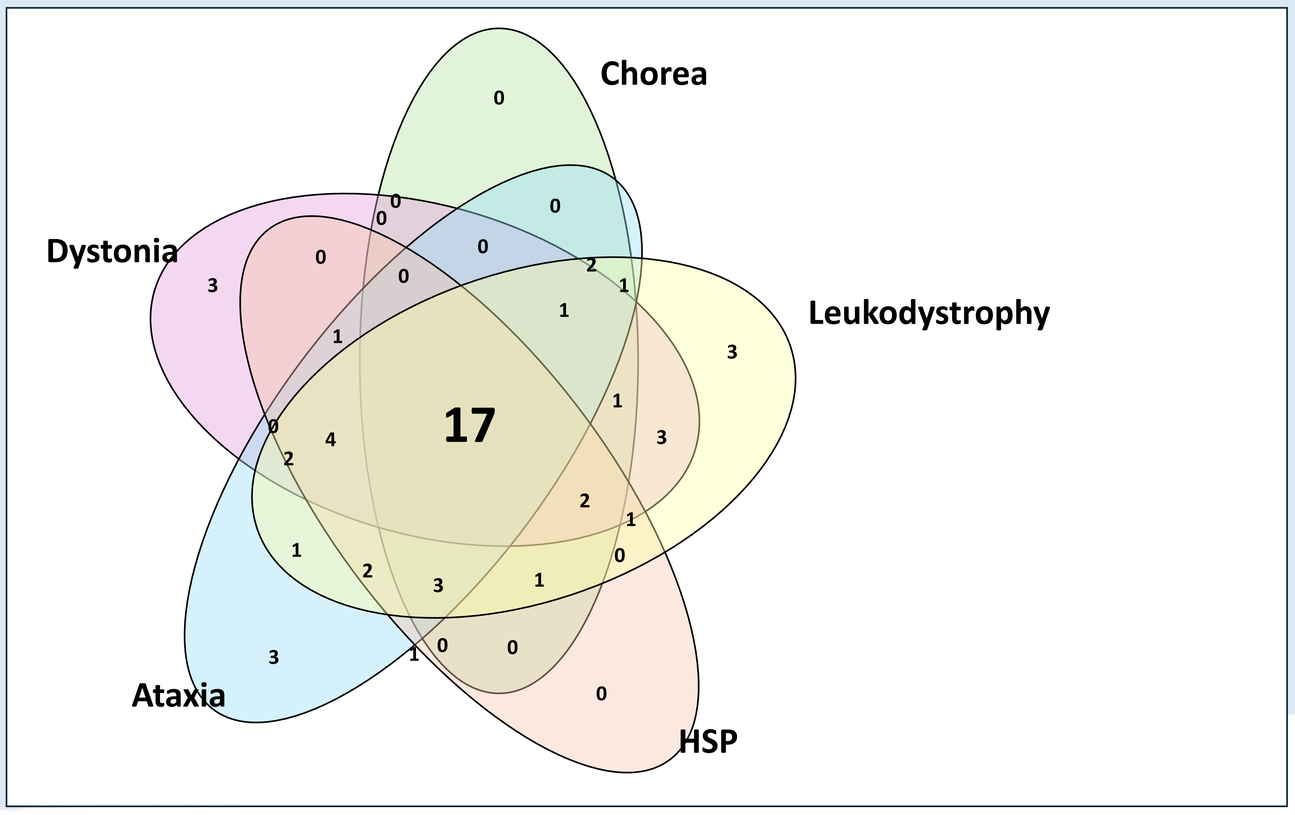


Figure 2.
Recommendations that reached consensus in each disease group.

## Outlook

Seventeen recommendations for clinical practice and scientific research met consensus in all disease groups, demonstrating their central role in transitional care. Recommendations cover specific fields including transition Procedure, Education, Documentation, Legal and Economic issues, External resources, and research Clinical tools. This study offers a set of core principles that could represent valid guidance for the transition process across rare neurological diseases. Additional emphasis should be placed on disease-specific issues and for a process personalized to the specific patient.

[1]Nanetti L, Kearney M, Boesch S, et al. Child-to-adult transition: a survey of current practices within the European Reference Network for Rare Neurological Diseases (ERN-RND). *Neurol Sci*. 2024;45(3):1007-1016. doi:10.1007/s10072-023-07101-3.

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